

# MitoBench: An interactive visual workbench for population genetics on mitochondrial DNA



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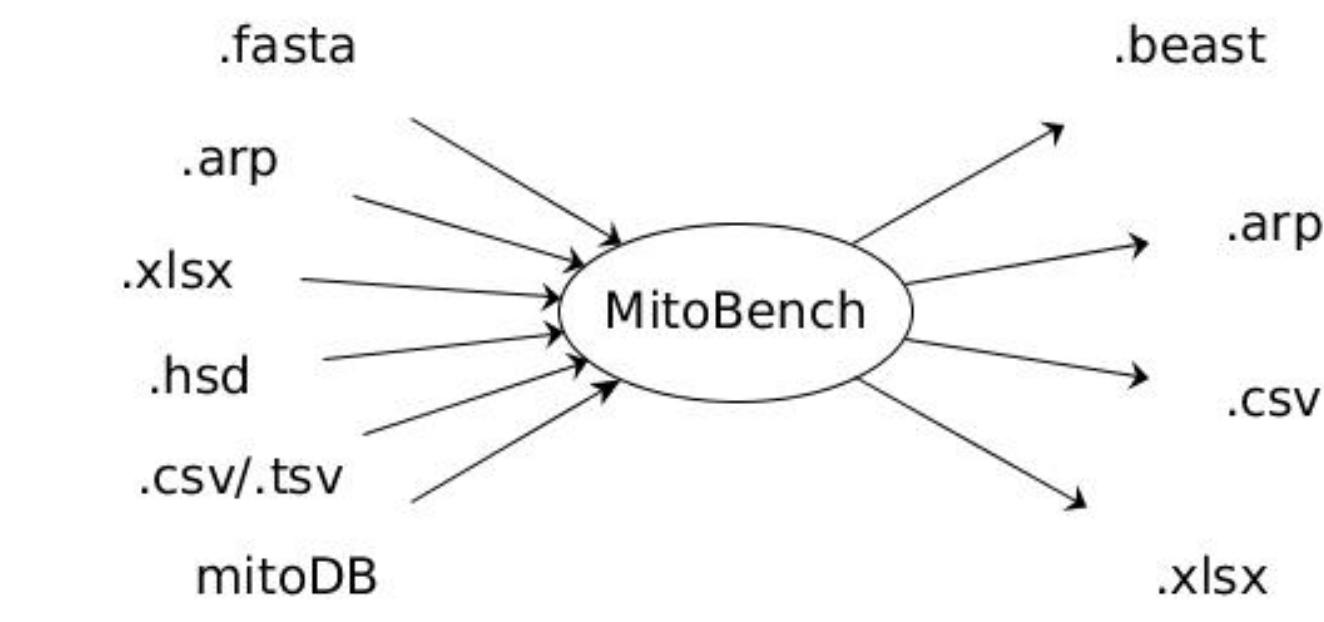
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## Introduction

Despite the availability of modern next generation sequencing technologies and therefore nuclear human genomes, the sequencing and analysis of mitochondrial DNA (mtDNA) is still common. Especially in the research field of ancient DNA and the context of population genetics, mtDNA is often the only proxy available to study extinct populations and their relationship with modern populations. As a consequence, many population genetic studies rely on the analysis of mtDNA. A plethora of methods for the analysis of mtDNA exist, that address questions in population genetics, phylogeny and others. However, these tools typically rely on different file formats and often require manual interaction with the data for downstream analysis. Ultimately, these steps can be cumbersome, especially for non-bioinformaticians, resulting in an increased risk of errors during the analysis. To tackle these issues, we present MitoBench, a workbench to interactively analyze and visualize mitochondrial genomes with a focus on population genetics. The graphical user interface is kept simple, to accommodate even users without further prior knowledge on computational methods. Furthermore, it shows additional information such as metadata and statistics. Currently, MitoBench offers automatic file conversion tools to connect the workbench with existing analysis methods such as BEAST, Arlequin and others.

## MitoBench

### Data import and export



### Data representation

ID	MTSequence	Haplogroup	Time Period	continent	country	country_region	culture_type	dating_lower
KJ2880	GATCA...	T1a2	Pre-Ptolemaic Period	Africa	Egypt	Beni Suef	Ancient	469
egyp1.5A129	GATCA...	X1c	Egypt Modern	Africa	Egypt	Other	Modern	2015
egyp1.6A08	GATCA...	H3w	Egypt Modern	Africa	Egypt	Other	Modern	2015
egyp1.305966	GATCA...	L3Q2a1	Ethiopia Modern	Africa	Ethiopia	Other	Modern	2015
egyp1.6A03	GATCA...	R0a3	Egypt Modern	Africa	Egypt	Other	Modern	2015
egyp1.305983	GATCA...	M1a5	Ethiopia Modern	Africa	Ethiopia	Other	Modern	2015
egyp1.305982	GATCA...	R0a2	Ethiopia Modern	Africa	Ethiopia	Other	Modern	2015
egyp1.306038	GATCA...	R0a2b	Ethiopia Modern	Africa	Ethiopia	Other	Modern	2015
egyp1.306032	GATCA...	L0P2b	Ethiopia Modern	Africa	Ethiopia	Other	Modern	2015
egyp1.306031	GATCA...	L3b2	Ethiopia Modern	Africa	Ethiopia	Other	Modern	2015
egyp1.306030	GATCA...	L2a1d1	Ethiopia Modern	Africa	Ethiopia	Other	Modern	2015
egyp1.7A139	GATCA...	R0b	Egypt Modern	Africa	Egypt	Other	Modern	2015
KJ2879	GATCA...	U3b	Roman Period	Africa	Egypt	Beni Suef	Ancient	2015
KJ2878	GATCA...	T1a7	Ptolemaic Period	Africa	Egypt	Beni Suef	Ancient	235
egyp1.8A134	GATCA...	H20c	Egypt Modern	Africa	Egypt	Other	Modern	2015

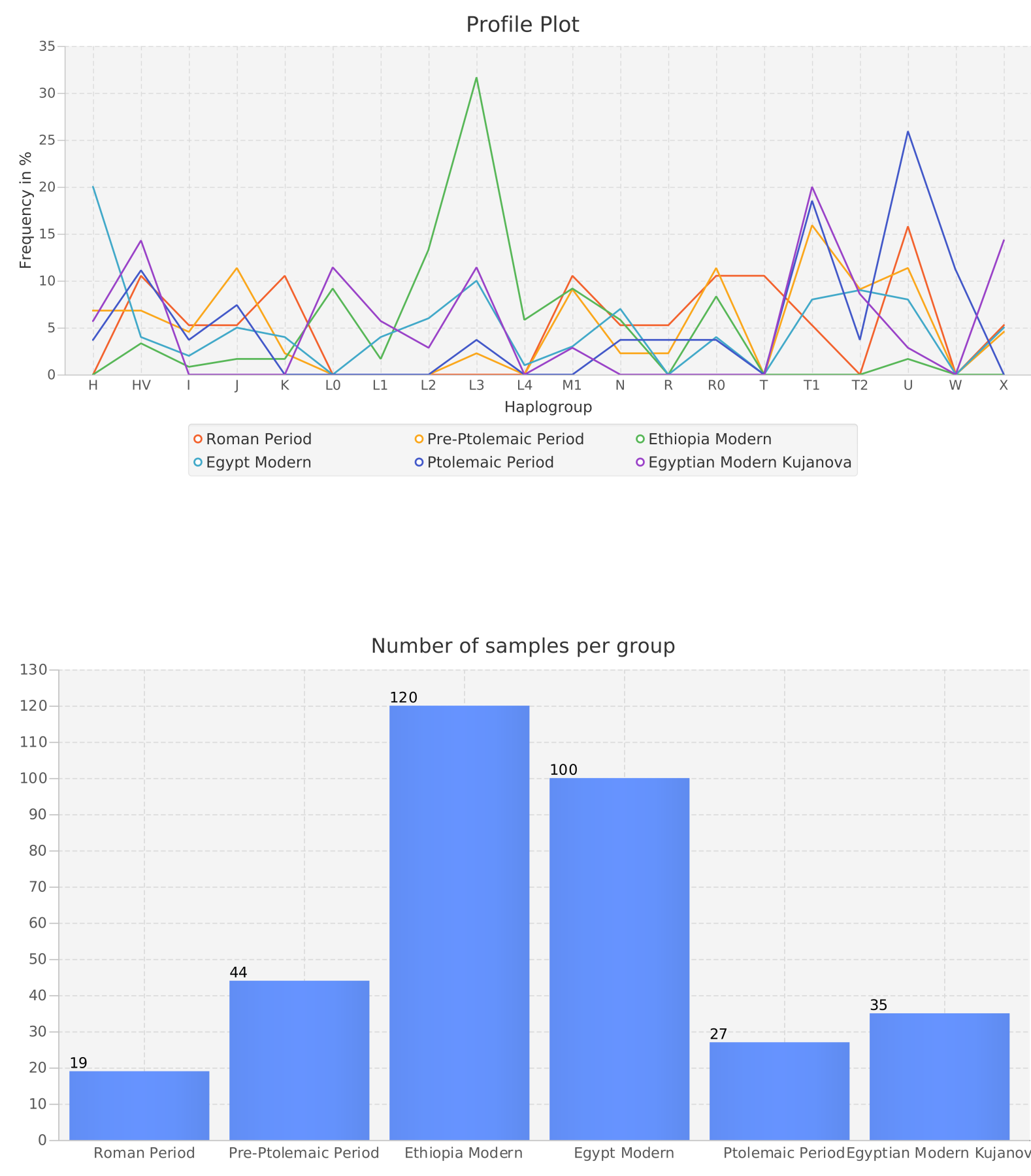
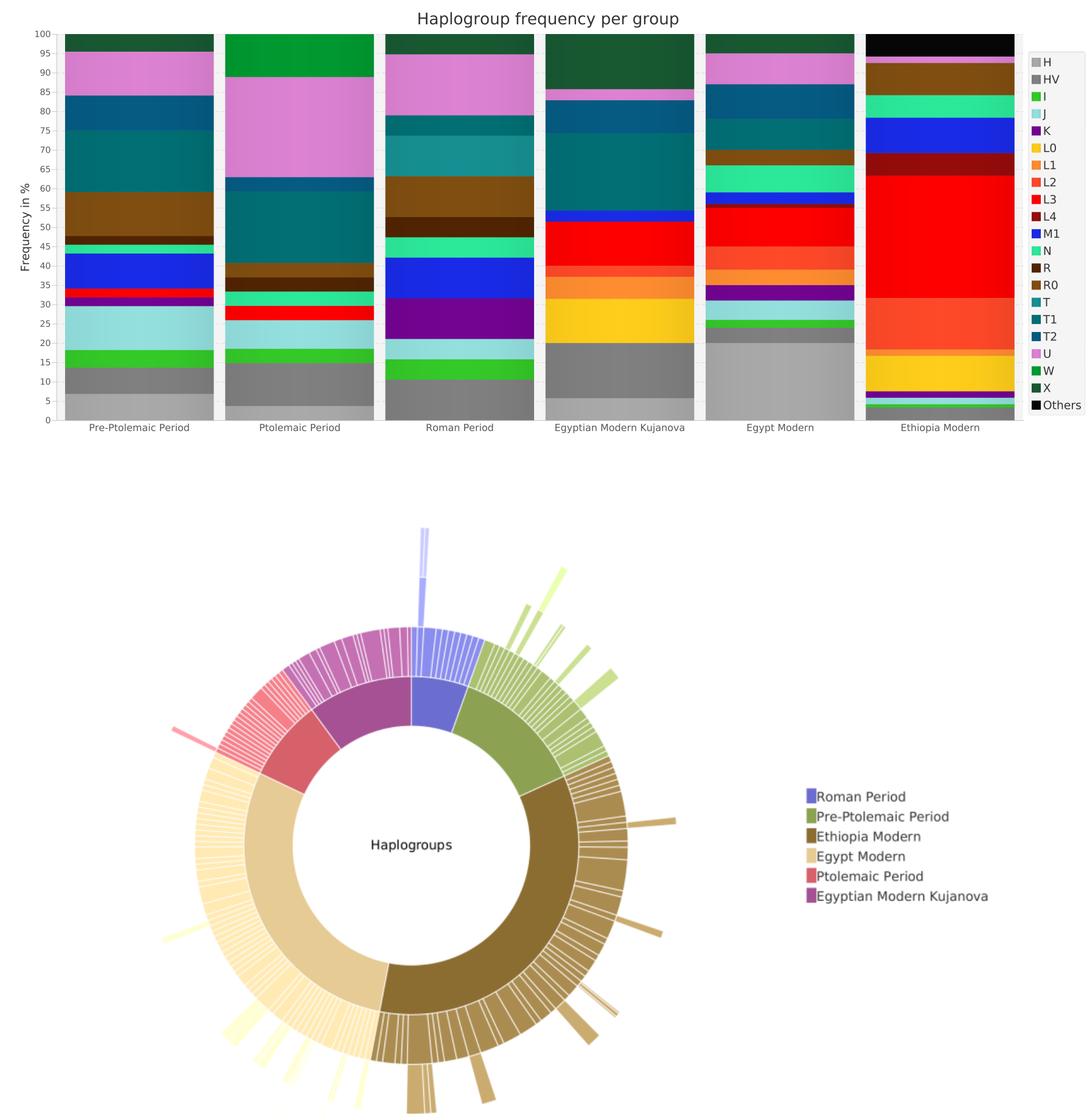
### Data grouping

- interested in behaviour of different groups  
→ Group data by feature
- internal grouping (columns not sorted)

### Data filtering / Statistics

- Haplogroup filtering / frequencies
- Mutation filtering / frequencies

### Data visualizations



## MitoDB

- Web-Frontend with Vaadin Java Framework
- Backend with PostgreSQL, providing sequence information and meta-data (SQL)
- Curated data upload with metadata
- Retrieval possible through WebUI and mitoBench

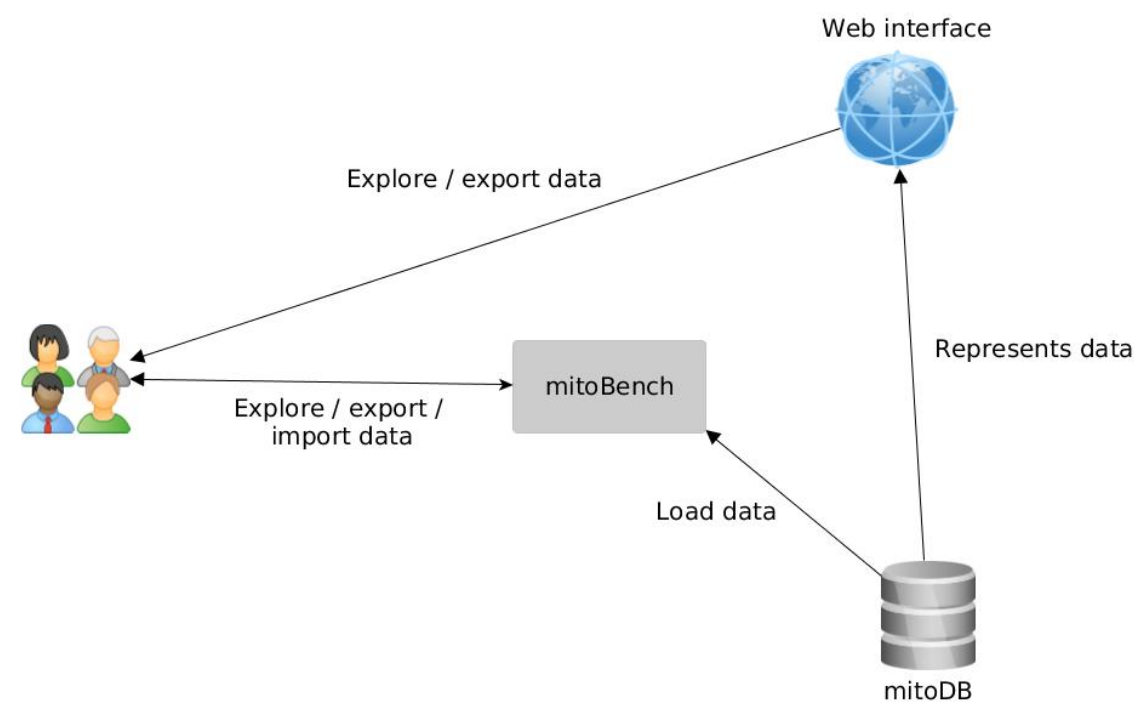
## Conclusion

### MitoBench

- Easy file handling and file conversion
- Combination, manipulation and visualization of mitochondrial data
- Data filtering
- Statistics

### MitoDB

- Collection of mitochondrial data plus meta information
- Curated database
- accessible via mitoBench and web interface



## References

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